



TAZ gene

tafazzin

Normal Function

The *TAZ* gene provides instructions for producing a protein called tafazzin. Several different versions (isoforms) of the tafazzin protein are produced from the *TAZ* gene. Most isoforms are found in all tissues, but some are found only in certain types of cells. Tafazzin is located in structures called mitochondria, which are the energy-producing centers of cells. Tafazzin is involved in altering a fat (lipid) called cardiolipin, which plays critical roles in the mitochondrial inner membrane. Tafazzin adds a fatty acid called linoleic acid to the cardiolipin molecule, which enables cardiolipin to perform its functions. Cardiolipin is necessary for maintaining mitochondrial shape, energy production, and protein transport within cells.

Health Conditions Related to Genetic Changes

Barth syndrome

More than 160 mutations in the *TAZ* gene have been found to cause Barth syndrome. This rare condition occurs almost exclusively in males and is characterized by an enlarged and weakened heart (dilated cardiomyopathy), muscle weakness, recurrent infections, and short stature. *TAZ* gene mutations that cause Barth syndrome result in the production of tafazzin proteins with little or no function. As a result, linoleic acid is not added to cardiolipin, which causes problems with normal mitochondrial shape and functions such as energy production and protein transport. Tissues with high energy demands, such as the heart and other muscles, are most susceptible to cell death due to reduced energy production in mitochondria. Additionally, affected white blood cells have abnormally shaped mitochondria, which could impair their ability to grow (proliferate) and mature (differentiate), leading to a weakened immune system and recurrent infections. Dysfunctional mitochondria likely lead to other signs and symptoms of Barth syndrome.

familial dilated cardiomyopathy

other disorders

Some mutations in the *TAZ* gene cause dilated cardiomyopathy without the other features of Barth syndrome (described above). Dilated cardiomyopathy is a condition in which the heart becomes weakened and enlarged and cannot pump blood efficiently, often resulting in heart failure. The decreased blood flow can lead to

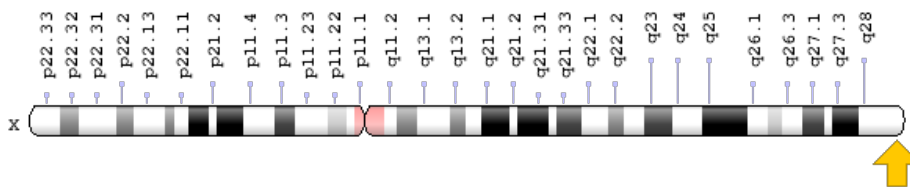
swelling in the legs and abdomen, fluid in the lungs, and an increased risk of blood clots.

Mutations in the *TAZ* gene can also cause a heart condition called isolated noncompaction of left ventricular myocardium (INVM). This condition occurs when the lower left chamber of the heart (left ventricle) does not develop correctly. In INVM, the heart muscle is weakened and cannot pump blood efficiently. Abnormal heart rhythms (arrhythmias) can also occur. INVM frequently causes heart failure.

Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 154,411,406 to 154,421,726 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BTHS
- CMD3A
- EFE
- EFE2
- G4.5
- LVNCX
- tafazzin (cardiomyopathy, dilated 3A (X-linked); endocardial fibroelastosis 2; Barth syndrome)
- TAZ_HUMAN
- XAP-2

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Mitochondria Import Most of Their Lipids; Chloroplasts Make Most of Theirs
<https://www.ncbi.nlm.nih.gov/books/NBK26924/#A2634>
- Molecular Biology of the Cell (fourth edition, 2002): The Mitochondrion Contains an Outer Membrane, an Inner Membrane, and Two Internal Compartments
<https://www.ncbi.nlm.nih.gov/books/NBK26894/#A2499>

GeneReviews

- Dilated Cardiomyopathy Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1309>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TAZ%5BTIAB%5D%29+OR+%28tafazzin%5BTIAB%5D%29+OR+%28Barth+syndrome%5BTIAB%5D%29%29+OR+%28%28BTHS%5BTIAB%5D%29+OR+%28CMD3A%5BTIAB%5D%29+OR+%28EFE%5BTIAB%5D%29+OR+%28EFE2%5BTIAB%5D%29+OR+%28G4.5%5BTIAB%5D%29+OR+%28tafazzin%5BTIAB%5D%29+OR+%28XAP-2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- TAFAZZIN
<http://omim.org/entry/300394>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TAZ.html
- Barth Syndrome Foundation: TAZ Gene Mutation Database
<https://www.barthsyndrome.org/science--medicine/human-taz-gene-variants-database>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TAZ%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11577

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6901>
- UniProt
<http://www.uniprot.org/uniprot/Q16635>

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